立法會秘書處
資料研究組

資料摘要
選定地方的罕見疾病政策

IN07/16-17

1. 引言

1.1 罕見疾病泛指多種患病率偏低的疾病，[1] 雖然個別罕見疾病的病例甚少，但若綜合全球逾 7 000 項罕見疾病計算，全球每 15 人即有 1 人罹患罕見疾病。[2] 罕見疾病涵蓋大部分先天遺傳疾病，此外，一些非常罕見的傳染病，自體免疫疾病及罕見癌症也屬於罕見疾病。[3]

1.2 罕見疾病通常是嚴重的慢性疾病，並且可能致命。罕見疾病種類紛繁，而醫學界對罕見疾病的認知不多，相關培訓亦不足，加上每項罕見疾病的患者為數甚少且分散各地，致使罕見疾病藥物(俗稱"孤兒藥")的研發工作成本既高，風險亦大，醫學界在應對罕見疾病方面，可謂挑戰重重。罕見疾病患者可能因為遲遲未能確診患病、無藥可治，及／或無力負擔昂貴的藥物和治療而延誤就醫。這些患者及其家人不但身心承受巨大壓力，經濟負擔亦十分沉重。

1.3 衞生事務委員會在 2016 年 12 月 19 日的會議上要求資料研究組研究海外地方對罕見疾病患者的支援政策，以便委員討論此議題。本資料摘要的研究對象包括美國、歐洲聯盟(下稱"歐盟")、日本及台灣；這些地方多年來致力制訂完善的醫療政策，以應對罕見疾病患者所面對的種種問題。下文各段概述香港及上述海外地方所制訂的罕見疾病政策。比較選定海外地方罕見疾病政策特點的摘要表載列於**附表**，隨後的**附錄 I 至附錄 IV** 則詳述各地政策，包括當地所採納的罕見疾病定義、其政策框架、相關法例、為支援患者所提供的醫療及社會服務，以及就診斷及治療患者方面的新近發展情況。**附錄 I 至附錄 IV** 只備英文本。

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1 罕見疾病亦稱為"孤兒病"，因為患者人數不多，藥廠在無利可圖的情況下缺乏誘因開發相關藥物。

2 請參閱 De Vrueh, R. et al (2013)，Shafie, A. et al (2016) and Song, P. et al (2012)。

3 請參閱 De Vrueh, R. et al (2013)。

2. 香港的罕見疾病政策

2.1 在香港，衞生署的醫學遺傳科為可能受遺傳病影響的家庭提供臨床診斷、輔導及預防服務。同時，醫院管理局亦為遺傳病患者提供醫療服務。此外，衞生署和醫院管理局於 2015 年 10 月開展"初生嬰兒代謝病篩查先導計劃"，以預防和減少由初生嬰兒代謝病而引起的嚴重健康問題。於 2016 年 4 月，該先導計劃所涵蓋的先天性代謝病數目由 21 項增至 24 項。

2.2 自 2008-2009 年度起，政府亦資助符合特定臨床用藥準則的 6 種溶小體儲積症[4] 患者接受酵素替代療法。最近，政府計劃透過關愛基金為特定罕見疾病(例如陣發性血紅素尿症)的合資格患者提供藥物資助。

2.3 政府雖已提供上述服務，但仍未就罕見疾病訂定任何正式定義，亦未有制訂任何具體政策，為罕見疾病患者提供支援。此外，部分主要持份者(尤其是病人組織)批評，從下列情況可見，政府未有為罕見疾病患者提供足夠支援：(a)罕見疾病的診斷需時漫長；(b)治療費用高昂，但只有少數患者獲提供藥物資助；(c)缺乏完整的病人資料庫，建立有關資料庫有助為患者提供具實證基礎的治療；及(d)未有提供足夠的社會服務以支援患者及其照顧者。[5]

3. 選定地方的罕見疾病政策

3.1 美國、歐盟、日本及台灣多年來致力制訂政策，以應對罕見疾病患者所面對的種種問題。這些地方除了清楚界定何謂罕見疾病外，亦制訂了指定罕見疾病藥物的制度，從而鼓勵藥廠開發治療罕見疾病的藥物。這些選定地方亦在各自的政策框架下推行其他措施，為罕見疾病患者提供醫療及／或社會服務。

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[4] 該 6 種溶小體儲積症包括高球氏症、龐貝氏症、一型／二型／六型黏多醣症，以及法柏氏症。截至 2015 年 12 月，醫院管理局已為 24 名溶小體儲積症患者提供酵素替代療法。

[5] 請參閱 Minutes of Meeting of the Panel on Health Services of the Legislative Council (2014)及 Hong Kong Alliance for Rare Diseases (2016)。

罕見疾病的定義

3.2 是次研究所涵蓋的所有海外地方皆按照疾病的患病率界定罕見疾病，但患病率的計算準則卻各有不同。美國按患者總數計算疾病患病率，國內患者少於 20 萬人的病症列作罕見疾病；歐盟及台灣以每 1 萬人中有多少名患者作為罕見疾病的界定準則(歐盟是每 1 萬人中少於 5 人，台灣則是每 1 萬人中少於 1 人)；日本則根據患者佔總人口的比例計算患病率，患者人數少於人口 0.1% 的病症即列為罕見疾病。此外，日本和台灣在界定某病症是否屬於罕見疾病時，亦會考慮其他準則，例如該病症是否難以診斷及治療。

指定罕見疾病藥物的制度

3.3 在是次研究所涵蓋的海外地方中，美國是首個制定特定法例的地方，藉以推動藥品業界研發治療罕見疾病的藥物。美國透過制定《孤兒藥品法》(Orphan Drug Act)訂立指定某藥物為罕見疾病藥物的準則，同時提供誘因，並在研發及申請審批藥物的過程中向藥廠提供協助，以鼓勵藥廠研發該等藥物。其後，日本、歐盟及台灣相繼通過法例，設立類似的制度，並向藥廠提供誘因，以解決罕見疾病"無藥可醫、無方可治"的困境。是次研究所涵蓋的海外地方向研發罕見疾病藥物的藥廠所提供的誘因包括：可就研究開支申請財政資助及稅務減免、精簡審批程序令藥物得以盡快推出市場，以及在藥物准予推出市場後享有一定年期的市場專賣權。

3.4 在是次研究所涵蓋的所有海外地方，當地在設立指定罕見疾病藥物的制度後，藥品業界均加強推展相關研發工作，而市場上亦有更多獲認可／經審批的相關藥物可供出售，以治療罕見疾病。舉例而言，在 2000 年至 2016 年期間，經歐盟指定的罕見疾病藥物共 1 805 項，當中 128 項獲批准在市場銷售，以治療 101 項罕見疾病。美國的情況相若，截至 2017 年 2 月底，獲指定的罕見疾病藥物數目高達 4 023 項。自 1983 年至今，在指定罕見疾病藥物的制度下，藥品業界已成功研發並獲准在市場銷售超過 575 項治療罕見疾病的藥物。相比在 1973 年至 1983 年間，藥品業界在市場推出的相關藥物少於 10 項。

為支援罕見疾病患者而推行的其他措施

3.5 在是次研究所涵蓋的所有海外地方，各地均推行下列措施，以促進公眾對罕見疾病的認識，以及加強及早識別和防治罕見疾病的工作：(a)透過設立網上資訊中心以提供有關罕見疾病的資訊；(b)推行新生嬰兒篩查計劃；(c)設立病人資料庫，以分享資料作治理病人和研究之用；及(d)投放資源以進行有關罕見疾病的研發工作。此外，日本和台灣均在其政策框架下採取具體措施，應對罕見疾病患者對社會服務的需要。尤其值得一提的是，兩地均將殘疾人士(persons with disabilities)(日本稱為"障害者"，台灣稱為"身心障礙者")的定義擴闊，使之涵蓋罕見疾病患者，並為他們提供相關法例所訂明的社會服務。

表 —— 選定地方的罕見疾病政策

	美國	歐洲聯盟	日本	台灣
罕見疾病的定義及普遍程度				
按照患病率界定何謂罕見疾病	• 患者人數少於 20 萬人。	• 在 1 萬人中少於 5 人。	• 患者人數少於全國人口 0.1%。	• 在 1 萬人中少於 1 人 (少於 0.01%)。
在指定某病症屬罕見疾病時考慮的其他準則	• 並無指明。	• 並無指明。	• 予以考慮的其他準則包括： (a) 病因不明； (b) 並無有效的治療方法； (c) 治療期漫長；及 (d) 病症有客觀的診斷標準。	• 予以考慮的其他準則包括： (a) 先天遺傳；及／或 (b) 難以診斷及治療。
當地人口罹患的罕見疾病數目／指定罕見疾病數目	• 約 7 000 項罕見疾病。	• 約 5 000 至 8 000 項罕見疾病。	• 306 項指定難治／罕見疾病。	• 約 210 項指定罕見疾病。
罕見疾病患者人數	• 在美國，罕見疾病患者人數介乎 2 500 萬至 3 000 萬人。	• 在歐洲聯盟，罕見疾病患者人數約為 3 000 萬人。	• 截至 2015 年年底，在日本的指定難治／罕見疾病患者人數為 943 460 人。	• 在 2015 年，在台灣的指定罕見疾病患者人數為 7 625 人。
政策框架				
負責機關	• 美國衞生與公眾服務部 (United States Department of Health and Human Services)；美國食品藥品管理局 (United States Food and Drug Administration)；以及有關的州立機關。	• 歐洲藥物管理局 (European Medicines Agency)；及個別成員國的有關機關。	• 厚生勞動省。	• 衞生福利部。

表 —— 選定地方的罕見疾病政策(續)

	美國	歐洲聯盟	日本	台灣
政策框架(續)				
相關法例	• 於 1983 年制定的《孤兒藥品法》(Orphan Drug Act)，以及於 2002 年制定的《罕見疾病法》(Rare Diseases Act)。	• 《歐洲聯盟孤兒藥品規例》(European Union Regulation on Orphan Medicinal Products)。	• 《藥事法》及《難治／罕見疾病患者醫療及社會支援法》。	• 《罕見疾病防治及藥物法》。
政策範疇	• 推動罕見疾病藥物的研發工作。 • 支援關乎罕見疾病的研發工作。	• 推動罕見疾病藥物的研發工作。 • 支援各成員國，確保各成員國制訂兼具效益與效率的措施，以識別、預防、診斷及治療罕見疾病，並就罕見疾病進行研究。	• 研發有效的治療方法，並加強為罕見疾病患者提供的醫療及社會服務。 • 設立公平而一視同仁的資助機制。 • 加深市民對罕見疾病的了解。	• 改善對罕見疾病的認知、預防、診斷及治療。 • 加強為罕見疾病患者提供的醫療及社會服務。
指定罕見疾病藥物的制度				
將某藥物指定為罕見疾病藥物的準則	• 有關藥物主治的疾病：(a)在美國的患者少於 20 萬人；或(b)在美國的患者超過 20 萬人，但該藥物在美國的銷量不足以收回其研發成本。	• 該藥物擬治療的疾病屬致命疾病，而該疾病的患病率符合列為罕見疾病的準則；同時該疾病並無有效的治療方案。	• 有關藥物必須符合 3 項準則：(a)日本只有少於 5 萬名病人使用有關藥物；(b)適用於治療嚴重疾病，而且沒有替代藥物可供選擇；及(c)有科學理據支持需要研發該藥物。	• 該藥物主要適用於預防、診斷及治療指定的罕見疾病。

表 —— 選定地方的罕見疾病政策(續)

	美國	歐洲聯盟	日本	台灣
指定罕見疾病藥物的制度(續)				
提供財務誘因及協助，以促進罕見疾病藥物的研發工作	• 誘因包括財政資助／稅務減免、精簡審批程序令藥物得以盡快推出市場，以及為期7年的市場專賣權。	• 誘因包括為期10年的市場專賣權、研究資助，以及減收向當局申請批准在市場銷售有關藥物的費用。	• 誘因包括財政資助／稅務寬免、為期10年的市場專賣權，以及精簡審批程序令藥物得以盡快推出市場。	• 誘因包括為期10年的市場專賣權，以及病人可提出專案申請，在指定罕見疾病藥物獲准推出市場前申請使用有關藥物並向當局申請發還購置藥物的費用。
指定罕見疾病藥物數目	• 4 023項 (截至2017年2月)。	• 1 805項 (2000年至2016年)。	• 327項 (截至2014年1月)。	• 98項 (截至2017年1月)。
獲准推出市場銷售的指定罕見疾病藥物數目	• 超過575項 (自1983年至今)。	• 128項 (2000年至2016年)。	• 203項 (截至2014年1月)。	• 並無相關資料。
向使用罕見疾病藥物的病人發還藥費	• 藥費由(a)病人所參與的公營或私營保險計劃承保；及(b)病人付出部分費用。	• 按照個別成員國的醫療融資制度及發還安排發還藥費予病人。	• 根據醫療保險制度，病人使用獲准在市場銷售的罕見疾病藥物可獲發還藥費。	• 病人可就使用經中央健康保險署批核的用藥清單上的罕見疾病藥物申請發還藥費。 • 若在使用有關藥物前提出申請並獲批准，可就不在清單上的藥物申請發還藥費。

表 —— 選定地方的罕見疾病政策(續)

	美國	歐洲聯盟	日本	台灣
為診斷及治療罕見疾病患者而推行的其他支援措施				
為加深主要持份者對罕見疾病的認知及認識而提供有關資訊	• 由基因與罕見疾病資訊中心(Genetic and Rare Diseases Information Center)提供淺白易明的最新資訊。	• 透過 Orphanet 入門網站提供全面及最新的資訊。	• 透過網上資源中心(即日本難病情報中心)提供相關資訊。	• 衛生福利部透過特定網站及各項公共教育計劃提供有關資訊。
為及早識別罕見疾病而採取的措施	• 個別州份各自推行新生嬰兒篩查計劃。	• 個別成員國各自推行新生嬰兒篩查計劃。	• 沒有在政府的政策框架下訂明。	• 新生嬰兒篩查計劃涵蓋 11 項先天性代謝疾病。
為協助罕見疾病患者獲取醫療服務而採取的措施	• 於 2010 年通過《病患保護及可負擔醫療法》(Patient Protection and Affordable Care Act)，以廢除一些針對罕見疾病患者的歧視性醫療保險條款。	• 視乎個別成員國的政策框架而定。	• 病人只須分擔兩成醫療費用，分擔費用設有每月上限，限額由厚生勞動省釐定。	• 在全民健康保險計劃之下，指定罕見疾病患者可獲發還八成醫療及醫藥費用。低收入病患者可獲發還全數費用。 • 如台灣當地沒有相關罕見疾病的診斷服務，患者可獲提供津貼，赴海外地方接受診斷服務。

表 —— 選定地方的罕見疾病政策(續)

	美國	歐洲聯盟	日本	台灣
為診斷及治療罕見疾病患者而推行的其他支援措施(續)				
為協助罕見疾病患者獲取社會服務而採取的措施	• 沒有在政府的政策框架下訂明。	• 沒有在政府的政策框架下訂明。	• 透過難治／罕見疾病諮詢及支援中心為患者提供社會服務。 • 擴闊"障害者"的定義，使之涵蓋大部分指定難治／罕見疾病患者，並為他們提供相關的社會服務。	• 擴闊"身心障礙者"的定義，使之涵蓋指定罕見疾病的患者，並為他們提供相關的社會服務。
設立病人資料庫／匯報制度	• 有。透過全球罕見疾病患者數據儲存庫(Global Rare Diseases Patient Registry Data Repository)，儲存由病人組織或研究人員設立的資料庫所載的病人資料。	• 有。各成員國各自設立病人資料庫。	• 有。	• 有。
投放資源以進行關乎罕見疾病的研發工作	• 有。國家衞生研究院(National Institutes of Health)轄下的罕見疾病研究辦公室(Office of Rare Diseases Research)負責推動有關罕見疾病的研發工作。	• 有。歐洲委員會資助歐洲各國與其他國家的機構聯手進行有關罕見疾病的研發工作。	• 有。厚生勞動省投放資源，以推展各種有關罕見疾病的研發項目。	• 有。衞生福利部提供誘因，鼓勵機構進行有關罕見疾病的研發工作。

Rare disease policy in the United States

A.I.1 In the US, the federal government enacted as early as in 1983 the Orphan Drug Act to support the medical treatment of rare disease patients. As discussed below, the Act provides for the establishment of an orphan drug designation system and the provision of various incentives to encourage research and development on orphan drugs for the treatment of rare diseases. In 2002, the federal government went further to enact the Rare Diseases Act to boost the research into rare diseases and the development of new treatments.

A.I.2 At the state level, individual states are responsible for setting and implementing their own medical care policies for the diagnosis and treatment of rare disease patients. According to the National Organization for Rare Disorders,[6] California has put in place a medical care policy with more comprehensive support to rare disease patients than other states.[7] Against this, the case of California is highlighted below to show how a comprehensive medical care policy could help a rare disease patient.

Orphan drug designation system

A.I.3 The US was the first place in the world to pass specific legislation designed to promote development of treatments for rare diseases, with provisions governing the designation of orphan drugs and granting of incentives to encourage pharmaceutical companies to develop orphan drugs. According to the Orphan Drug Act, the US Food and Drug Administration[8] will define a drug as an orphan drug if it treats a disease which: (a) affects less than 200 000 persons in the US; or (b) affects more than 200 000 persons in the US but the cost of developing and producing the drug is not expected to be recovered from drug sales in the country.

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[6] The National Organization for Rare Disorders is an independent, non-profit advocacy organization representing rare disease patients and their families in the US.

[7] See National Organization for Rare Disorders (2016a).

[8] The US Food and Drug Administration is a federal agency responsible for inspecting, testing, approving, and setting safety standards for foods and food additives, drugs, chemicals, cosmetics, and household and medical devices.

A.I.4 The Orphan Drug Act also provides for various incentives to promote the development of orphan drugs in the US. For example, the manufacturer of a designated orphan drug is entitled to (a) financial subsidies and tax credit on costs of clinical studies; (b) a seven-year period of marketing exclusivity following the grant of marketing approval to protect the drug from competition from similar products; and (c) a fast-track marketing approval procedure. Manufacturers have to obtain marketing approval from the US Food and Drug Administration before they can sell their orphan products in the market. Nonetheless, an orphan drug may be adopted for compassionate use before a marketing approval is granted under specified conditions.[9]

Access to medication of rare diseases

A.I.5 Under the health insurance system in the US, patients' medical and medication costs are covered by the public or private insurance schemes that they have enrolled in and co-payments of patients.[10] Previously, rare disease patients could not access healthcare coverage due to various discriminatory insurance practices. The passage of the Patient Protection and Affordable Care Act in 2010 successfully reformed these practices by forbidding insurers from discriminating against rare disease patients (such as denying coverage for patients having a pre-existing condition) and outlawing annual and lifetime coverage limits.[11]

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[9] The specified conditions are that: (a) the drug is intended to treat a serious or immediately life-threatening disease; (b) there is no satisfactory alternative treatment available; (c) the drug is already under investigation or trials have been completed; and (d) the pharmaceutical company is actively pursuing marketing approval.

[10] Eligible low-income persons and persons with disabilities are provided with free or low-cost medical benefits under the Medicaid programme jointly funded by the federal and state governments.

[11] The Patient Protection and Affordable Care Act has been opposed by some stakeholders as it has brought about issues such as increase in insurance costs from better insurance coverage. Currently, the federal government has been reviewing the health insurance system with a view to repealing the Act.

Other support measures

A.I.6 The federal government enacted the Rare Diseases Act in 2002 to empower the Office of Rare Diseases Research ("ORDR[12]") under the National Institutes of Health ("NIH[13]") to promote research and development on diagnostics and treatments for rare diseases. Pursuant to the Act, NIH set up the Genetic and Rare Diseases Information Center in 2002 to provide patients, their families and medical professionals access to current and easy-to-understand information about rare diseases. ORDR also set up a rare diseases clinical research network in 2009 to enhance collaboration and information sharing among research institutions in the US and other countries.

A.I.7 In addition, NIH developed the Global Rare Diseases Patient Registry Data Repository in 2010 to aggregate and store patient information from different registries set up by patient advocacy groups or researchers. The Repository enables stakeholders to access information about multiple rare diseases through a centralized source and facilitates research on rare diseases.

Rare disease policy in California

A.I.8 In California, the California Department of Public Health ("CDPH") has implemented a newborn screening programme to facilitate early identification and intervention of genetic and congenital disorders among babies.[14] At present, the screening programme covers some 80 disorders including metabolic, endocrine and hemoglobin disorders. In 2016, the progamme has further been enhanced after the enactment of a law requiring CDPH to expand the programme to include screening for any disease within two years after the disease is included in the list of conditions for screening recommended by the Advisory Committee on Heritable Disorders in Newborns and Children.[15]

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[12] ORDR was established in 1993 within the NIH Office of the Director. In 2012, ORDR became part of the National Center for Advancing Translational Sciences tasked to develop innovations in an effort to speed the delivery of new drugs, diagnostics and medical devices to patients.

[13] NIH is an agency under the Department of Health and Human Services responsible for providing leadership and direction to research programmes to improve the health of the public. NIH comprises 27 institutes and centres with each having its own specific research agenda.

[14] All the states in the US have implemented a newborn screening programme as a public health programme. However, the conditions screened under their respective programmes vary.

[15] The Advisory Committee is tasked to advise the Secretary of the Department of Health and Human Services on the development of newborn screening policies and programmes for reducing morbidity and mortality in newborns and children having heritable disorders.

A.I.9 The protection of patients, including those suffering from rare diseases has been enhanced after the recent passage of a law that limits patients' out-of-pocket payments for prescription drugs. Patients' access to appropriate drugs and treatments has also been enhanced by laws stipulating that (a) doctors can appeal against insurers' decision to adopt step therapy[16] for their patients, and (b) pharmacists must communicate to the patients and their doctors if they substitute a biologic drug with a lower cost non-identical alternative.

Recent developments on diagnosis and treatments of rare diseases

A.I.10 NIH estimates that between 25 million to 30 million persons in the US are affected by about 7 000 rare diseases.[17] The enactment of the Orphan Drug Act in 1983 has increased the number of designated orphan drugs to 4 023 at end-February 2017. The Act has also spurred the development of drugs for treating rare diseases, leading to the development and marketing of more than 575 orphan drugs and biologic products. In contrast, fewer than 10 orphan drugs were introduced in the market between 1973 and 1983. As for research on rare diseases, funding of the researches undertaken by NIH amounted to US$3.8 billion (HK$29.5 billion) in 2016, up from US$3.6 billion (HK$27.9 billion) in 2012.

A.I.11 Nonetheless, patient advocacy groups are concerned that approved orphan drug treatments are only available for less than 10% of rare diseases affecting patients in the US.[18] Besides, they are discontented with the high disparity among states in terms of the coverage of the state newborn screening programmes and the protection provided for patients under the health insurance systems. They have lobbied the state governments to enhance support and protection of rare disease patients.

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[16] Insurers may seek to control costs with the use of step therapy requiring insured patients to take one or more different lower-cost medications before getting the one that their doctors originally recommended.

[17] See National Institutes of Health (2016).

[18] See National Organization for Rare Disorders (2016b).

Rare disease policy in the European Union

A.II.1 In the EU, rare diseases were identified, for the first time, as a "priority field" for public health action in 1993. In 1999, rare diseases were further classified as a "priority area" for community action in the context of public health and the first programme of community action was adopted for the period between 1999 and 2003. The programme focused on improving knowledge and facilitating access to information about rare diseases for enhancing health protection of patients. Under the programme, rare diseases were defined as any disease affecting fewer than five in 10 000 persons in the EU.

A.II.2 In 2000, the European Commission[19] established an orphan drug designation system pursuant to the EU Regulation on Orphan Medicinal Products in order to promote the development of orphan drugs for treating rare diseases in the EU. In 2008, the European Commission set out an overall Community strategy for supporting member states in ensuring effective and efficient recognition, prevention, diagnosis and treatment of, and research on rare diseases in the EU. Member states have also been engaging in developing national plans to combat rare diseases since 2009.

Orphan drug destination system

A.II.3 Under the orphan drug designation system, the European Commission will consider to grant orphan drug designation to a medicine if it meets the following three criteria: (a) the medicine must be intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition; (b) the prevalence of the condition in the EU must be no more than five in 10 000 persons or it is unlikely that marketing of the medicine would generate sufficient returns to justify the investment needed for its development; and (c) no satisfactory method of diagnosis, prevention or treatment of the condition concerned has been authorized or, if such a method exists, the medicine will be of significant benefit to those affected by the condition.

[19] The European Commission is the executive arm of the EU and promotes its general interest.

A.II.4 Pharmaceutical companies developing designated orphan drugs are offered with incentives at the EU and individual member state levels to encourage their research and development activities. These incentives include: (a) scientific advice of the European Medicines Agency[20] at a reduced fee; (b) reduced fees for marketing authorization applications and other regulatory activities; (c) research grants; and (d) a 10-year period of marketing exclusivity after obtaining marketing authorization.

Access to medication of rare diseases

A.II.5 All designated orphan drugs are required to be assessed through a centralized marketing authorization procedure administered by the European Medicines Agency and granted authorization by the European Commission before they can be marketed in the EU.[21] Nonetheless, many member states have implemented compassionate use programmes enabling patients to access orphan drugs that are being considered for but not yet been granted marketing authorization under specified conditions.[22] The costs of orphan drugs incurred by patients are reimbursed in accordance with the healthcare financing systems and reimbursement arrangements put in place by individual member states.[23]

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[20] The European Medicines Agency is an agency of the EU responsible for the scientific evaluation, supervision and safety monitoring of medicines developed by pharmaceutical companies for use in the EU.

[21] Under the centralized authorization procedure, pharmaceutical companies can submit a single marketing authorization application to the European Medicines Agency and the authorization is valid across all EU member states once it is granted.

[22] For example, Germany allows patients with a seriously debilitating or life-threatening disease to access unauthorized orphan drugs if they cannot be treated satisfactorily with an authorized medicine.

[23] For example, the costs of using authorized orphan drugs are fully reimbursed by statutory health insurance in Germany.

Other support measures

A.II.6 Based on the overall Community strategy on rare diseases adopted in 2008, the European Commission has implemented a series of measures to pool resources across the EU member states to help patients and medical professionals share information and expertise across borders. These measures include: (a) funding operation of the Orphanet portal[24] to provide comprehensive and updated information on rare diseases and orphan drugs; (b) providing supporting tools to individual member states to help them develop their national plans on rare diseases; (c) funding the collaborative research projects on the causes of rare diseases, and on preventive, diagnostic and therapeutic interventions; (d) supporting the development of patient registries by individual member states to facilitate clinical research on rare diseases and patient care planning, and developing a European platform on rare diseases registration; and (e) evaluating the current practices of newborn screening for rare diseases in individual member states and exploring potential areas of collaboration among member states in this field.

Recent developments on diagnosis and treatments of rare diseases

A.II.7 At present, it is estimated that there are 5 000 to 8 000 rare diseases affecting about 30 million people in the EU.[25] According to the European Commission, the implementation of the overall Community strategy on rare diseases has fostered co-operation and exchange in experiences among the EU member states and other stakeholders. The strategy has also supported the EU member states to develop their national plans for rare diseases. As at February 2017, 22 out of the 28 member states have put in place dedicated plans to combat rare diseases. Besides, the European Commission has funded some 120 collaborative research projects relating to rare diseases between 2007 and 2013 with a total budget of over €620 million (HK$6,386 million).

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[24] Orphanet is a free multilingual portal administered by the European Commission on rare diseases and orphan drugs. It contains information on more than 5 000 rare diseases and aims to improve the quality of medical care and provide specialized services for the rare disease community.

[25] See European Medicines Agency (2017a).

A.II.8 Between 2000 and 2016, the European Commission has designated 1 805 orphan drugs, and granted marketing authorizations for 128 orphan drugs for treating 101 conditions. Nonetheless, there has been disparity in patients' access to authorized orphan drugs among the EU member states as they have set different pricing and reimbursement mechanisms. Member states have also faced challenge in providing affordable and sustainable access to drugs for rare disease patients as possible treatments are scarce and expensive.

Rare disease policy in Japan

A.III.1 In Japan, rare diseases are categorized as intractable diseases characterized by (a) a lack of an identifiable cause and a clearly established treatment; (b) having a considerable high risk of disability; and (c) posing heavy economic and psychological burdens on the patients and their families. The Japanese government first introduced a dedicated policy to combat intractable/rare diseases in 1972. Under the policy, the Specified Disease Treatment Research Programme was implemented to promote research on intractable/rare diseases and provide financial subsidy to patients. Eligible intractable/rare disease patients who had participated in the research programme were provided with subsidy on their medical costs.[26]

A.III.2 Japan has seen improvements in the medical treatments for patients suffering from intractable/rare diseases and advancement in related research activities since the introduction of the first policy on intractable/rare diseases in 1972. Nonetheless, in order to further enhance the support provided for intractable/rare disease patients, the Japanese government conducted a review on its intractable/rare disease policy in 2011 and subsequently enacted the Act on Medical Care and Social Supports for Patients with Intractable/Rare Diseases in 2014. The Act lays down the framework for provision of comprehensive support for intractable/rare disease patients in three major areas: (a) development of effective treatments for intractable/rare diseases and improvement of care for those affected; (b) establishment of a fair and consistent subsidization mechanism for patients' medical expenses; and (c) implementation of measures to enhance public understanding of intractable/rare diseases and encourage social participation of patients.

[26] For patients participating in the Specified Disease Treatment Research Programme, the co-payment bore by the patients was capped at a limit specified by the Ministry of Health, Labour and Welfare. Patients with low income and those with severe medical conditions were fully subsidized on their co-payment.

Orphan drug/medical device designation system

A.III.3 To encourage the research and development of life-saving but generally unprofitable drugs and medical devices for the treatment of intractable/rare diseases, the Japanese government established an orphan drug/medical device designation system in 1993 pursuant to an amendment of the Pharmaceutical Affairs Act. Under the system, the Ministry of Health, Labour and Welfare ("MHLW") will consider to designate a product as an orphan drug or medical device based on the following three criteria: (a) the product will be used by less than 50 000 patients; (b) it should be indicated for the treatment of serious diseases and there is no appropriate alternative available or is clinically superior to products available in the market; and (c) there should be a scientific rationale for using the product for the target disease and the development plan should be appropriate.

A.III.4 MHLW offers incentives to institutions developing designated orphan drugs/medical devices to support their research and development activities. These incentives include: (a) financial aids such as subsidy and tax relief for research expenses; (b) guidance and consultation from the Pharmaceuticals and Medical Devices Agency[27] at a lower fee; (c) a fast-track marketing approval process;[28] and (d) a 10-year period of marketing exclusivity. The costs incurred by intractable/rare disease patients on orphan drugs granted with marketing approval can be reimbursed under the health insurance system of Japan at a price set by MHLW.[29]

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[27] The Pharmaceuticals and Medical Devices Agency is the government organization in Japan in charge of reviewing drugs and medical devices, overseeing post-market safety, and providing relief for adverse health effects. The Agency charges the pharmaceutical companies for the advice and consultation provided.

[28] Institutions intending to manufacture or sell drugs or medical devices in Japan are required to obtain an approval from MHLW for each product. The products will be examined on their efficacy and safety in the approval process.

[29] The fees for medical care and medications charged by medical institutions and pharmacies on patients insured under the national health insurance system are set by MHLW based on the recommendations of the Central Social Insurance Medical Council.

A.III.5 In 2015, MHLW implemented a new scheme for subsidizing the medical fees of patients suffering from designated intractable/rare diseases. Designated intractable/rare diseases are defined as those diseases that meet the following criteria: (a) affecting less than 0.1% of the population in Japan; (b) causes not being identified; (c) lacking effective treatments; (d) requiring long-term treatments; and (e) existence of objective diagnostic criteria. The number of intractable/rare diseases covered under the new scheme increased from 56 to 306 by July 2015. Patients are only required to bear 20% of the medical costs as co-payment, capped at a monthly limit set by MHLW. The figure is lower than the 30% co-payment rate applicable to general patients covered under the health insurance system in Japan.

Other support measures

A.III.6 On medical care services, MHLW has improved the service provision by designating hospitals and doctors that are specialized in treating intractable/rare diseases for providing diagnoses and treatments for patients. Designated doctors are required to meet specified level of experience and training. For further improving the quality of medical care for patients, the local public health centres provide outreach and community care services to support these patients. MHLW also commits resources on research on diagnosis, prevention and treatment of intractable/rare diseases, and patients' quality of life.

A.III.7 In addition, MHLW has enhanced the national registry of designated intractable/rare diseases since 2015 to enable collection of data from designated doctors. The enhancement facilitates sharing of patient information among medical institutions and professionals for patient care planning and research purposes.[30]

[30] The national registry was established before 2000 and data had been collected by the local public health centres mainly for administration of the patient subsidy scheme and research on intractable/rare diseases.

A.III.8 As for social care services, MHLW has enhanced the capabilities of the local intractable/rare diseases consultation and support centres[31] in providing social care services for patients such as counselling and employment support services. These consultation and support centres team up with the local public health centres on providing coordinated social and medical care services for patients. More importantly, patients suffering from most designated intractable/rare diseases are included in the definition of persons with disabilities under the General Support for Persons with Disabilities Act. They are entitled to welfare services stipulated in the Act, which include nursing care services, community life support services, and training services for securing or sustaining employment.

A.III.9 In addition to the above, the Japan Intractable Diseases Information Center has been a major information source for intractable/rare disease patients and other key stakeholders (e.g. patients' families and medical professionals). This online resource centre was established by MHLW in 1997 in corporation with the Japan Intractable Diseases Research Foundation. MHLW has planned to further enhance the content of the Information Centre to promote public understanding of intractable/rare diseases.

Recent developments on diagnosis and treatments of rare diseases

A.III.10 As at end-2015, about 943 460 patients suffering from designated intractable/rare diseases were receiving financial subsidy for their medical costs under the new subsidization scheme set out in the Act on Medical Care and Social Supports for Patients with Intractable/Rare Diseases of 2014. The estimated cost of the subsidization scheme in 2015 was ¥222 billion (HK$14.2 billion). These compare with 855 061 patients and ¥133.5 billion (HK$10.6 billion) under the old scheme in 2013. Regarding the development of orphan drugs, MHLW had designated 327 products as orphan drugs as at January 2014, of which 203 products had been granted marketing approval. The majority of orphan drugs approved in Japan are for treating infectious diseases, haematological diseases, neuromuscular diseases, and diseases common in children and infants.

[31] MHLW has supported the local governments to set up intractable/rare diseases consultation and support centres since 2003 to provide social care services for intractable/rare disease patients.

Rare disease policy in Taiwan

A.IV.1 Prior to 2000, Taiwan's National Health Insurance programme[32] did not cover the drug and treatment costs incurred by rare disease patients unless the rare diseases had been defined as major illnesses. In order to enhance the support to rare disease patients, the Taiwanese government promulgated the Rare Disease and Orphan Drug Act ("Rare Disease Act") 《罕見疾病防治及藥物法》 in August 2000. The Act lays down a comprehensive framework to improve the awareness, prevention, diagnosis and treatment of rare diseases in Taiwan. The Taiwanese government aims to provide rare disease patients with easier access to drugs and life-sustaining nutritional supplements by promoting and ensuring the research and development, manufacturing and supply of these products.

Orphan drug designation system

A.IV.2 According to the Rare Disease Act, rare diseases are defined as diseases having a prevalence rate of lower than 1 in 10 000 persons or 0.01%, or diseases meeting other specified criteria such as having a genetic origin or being difficult to diagnose and treat. Applications for designation of rare diseases are reviewed by the Committee for the Review of Rare Diseases and Orphan Drugs ("the Review Committee") under the Ministry of Health and Welfare ("MOHW"). The Review Committee also considers applications for designation of orphan drugs which are defined as drugs having major indications for the prevention, diagnosis and treatment of rare diseases.

[32] The National Health Insurance programme is a mandatory social insurance programme to cover the medical and medication costs of the Taiwanese people. The programme is financed by premiums contributed by the insured, employers and the government.

A.IV.3 To encourage the development and supply of orphan drugs for treatment of rare diseases, MOHW offers the following incentives to importers and manufacturers of designated orphan drugs: (a) going through a simplified market approval procedure administered by the Taiwan Food and Drug Administration ("TFDA") under MOHW;[33] (b) reduction in registration fee; (c) granting a 10-year period of marketing exclusivity; and (d) allowing special application for usage and reimbursement prior to market approval. Besides, MOHW has introduced incentive schemes to encourage institutions to engage in research and development and other relevant activities that contribute to the prevention and control of rare diseases in Taiwan.

Access to medication of rare diseases

A.IV.4 Importers or manufacturers of designated orphan drugs have to apply for listing of the drugs by the National Health Insurance Administration ("NHIA")[34] before reimbursement of the drug costs borne by rare disease patients can be arranged. However, patients or medical institutions can apply for a permit to import a designated orphan drug without market approval or a non-designated orphan drug on an ad hoc basis.[35] For reimbursement of orphan drugs that are not on the NHIA reimbursement list, approval by the Review Committee has to be sought prior to usage of the drugs.

[33] According to the Pharmaceutical Affairs Act, drug importers or manufacturers are required to register their products with and obtain market approval from TFDA before they can sell or manufacture their products in Taiwan. Applicants for registration and market approval may be required to conduct local clinical trials of the drug where necessary.

[34] NHIA is an administrative agency under MOHW tasked to administer the National Health Insurance programme.

[35] The amount of drug allowed to be imported is limited to the usage amount required by the patient in two years.

A.IV.5 According to the Rare Disease Act, patients suffering from the designated rare diseases are entitled to reimbursement of up to 80% of the costs of diagnostic services, designated orphan drugs, treatments and supportive equipment, and 100% of costs of designated life-sustaining nutritional supplements[36] under the National Health Insurance programme. Patients with low income are provided with full reimbursement of the medical costs. MOHW also subsidizes patients to travel overseas for diagnosis of rare diseases or transfer specimens to overseas laboratories for testing in case the diagnoses or tests cannot be done locally. In addition, MOHW has set up an orphan drug and nutritional supplement supply centre to provide medical institutions with specific orphan drugs and life-sustaining nutritional supplements for emergency use.

A.IV.6 Apart from the support measures on treatment of rare diseases, MOHW has also certified 14 genetic consultation centres and some 30 genetic diagnostic laboratories to provide consultation and diagnostic services for rare disease patients. A central reporting system for rare diseases has been established to facilitate the provision of medical care services for patients, and enhance prevention and control of rare diseases.

Other support measures

A.IV.7 Since 2002, persons suffering from designated rare diseases are included under the definition of persons with disabilities and protected under the People with Disabilities Rights Protection Act《身心障礙者權益保障法》. Rare disease patients are entitled to specified social services and benefits such as social security benefits, employment assistance services, tax deduction and fare discount for public transport.

[36] Applications for designation of life-sustaining nutritional supplements which are defined as foods primarily suited for providing nutrients to rare disease patients are considered by the Review Committee.

A.IV.8 Meanwhile, MOHW has put in place various measures to promote the awareness and early diagnosis of rare diseases. In 2006, it expanded the newborn screening programme to cover 11 metabolism disorders to facilitate early identification and treatment of babies with genetic disorders.[37] Added to this, MOHW has set up a portal for providing information about rare diseases and diagnostic services available, as well as implementing a series of public education progammes since 2000 to raise public awareness of rare diseases and lower the barriers to receiving treatments among patients.

Recent developments on diagnosis and treatments of rare diseases

A.IV.9 According to MOHW, there were about 210 designated rare diseases and 98 designated orphan drugs in Taiwan at end-January 2017. Latest statistics published by NHIA reflected that there were 7 625 patients suffering from designated rare diseases in 2015, and the average drug cost incurred per patient was about NT$542,000 (HK$136,000). Nevertheless, the implementation of the Rare Disease Act in 2000 and the various support measures so provided have helped rare disease patients relieve their economic and psychological burdens.

A.IV.10 Nonetheless, as reimbursement of the costs of using designated orphan drugs can be arranged before the registration and market approval process is completed, NHIA has indicated that some drug suppliers are unwilling to complete the process, leading to incomplete information about the therapeutic and adverse effects of the drugs.

[37] The newborn screening programme was first introduced in 1985 and only covered five metabolism disorders.

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資料摘要為立法會議員及立法會轄下委員會而編製，它們並非法律或其他專業意見，亦不應以該等資料摘要作為上述意見。資料摘要的版權由立法會行政管理委員會("行政管理委員會")所擁有。行政管理委員會准許任何人士複製資料摘要作非商業用途，惟有關複製必須準確及不會對立法會構成負面影響，並須註明出處為立法會秘書處資料研究組，而且須將一份複製文本送交立法會圖書館備存。本資料摘要的文件編號為 IN07/16-17。